We claim:

1. A method for testing a sample for mutations in the BRCA1 gene comprising the steps of

(a) amplifying one or more exons or partial exons of BRCA1 to produce amplified fragments;

(b) determining the sizes and amounts of the amplified fragments and comparing the determined sizes or amounts to standard values for amplification of the same exons or partial exons of wild-type BRCA1 gene, wherein a difference in fragment size or amount is indicative of the presence of a mutation in the BRCA1 gene; and

(c) if no mutation is detected in the BRCA1 gene as a result of the determination of the sizes and amounts of the amplification fragments, determining the sequence of one or more exons of the BRCA1 gene.

2. The method according to claim 1, wherein at least exons 2, 11 and 20, or portions thereof, are amplified to produce amplification fragments.

3. The method according to claim 1, wherein two or more exons or partial exons are amplified in a multiplex amplification reaction to produce multiplex amplification products.

4. The method according to claim 3, wherein exons 1 and 21 of the BRCA1 gene are amplified in one multiplex amplification reaction.

5. The method according to claim 4, wherein exons 1 and 21 are amplified using the primers identified by Sequence ID Nos. 1, 2, 67 and 68, or primers complementary thereto.

6. The method according to claim 3, wherein exons 2, 5, 9 and 14 of the BRCA1 gene are amplified in one multiplex amplification reaction.

7. The method according to claim 6, wherein exons 2, 5, 9 and 14 are amplified using the primers identified by Sequence ID Nos. 3, 4, 9, 10, 17, 18, 53 and 54, or primers complementary thereto.

8. The method according to claim 3, wherein exons 3, 7 and 15 of the BRCA1 gene are amplified in one multiplex amplification reaction.

9. The method according to claim 8, wherein exons 3, 7, and 15 are amplified using the primers identified by Sequence ID Nos. 5, 6, 13, 14, 55, and 56, or primers complementary thereto.

10. The method according to claim 3, wherein exons 6, 10, 17 and 18 of the BRCA1 gene are amplified in one multiplex amplification reaction.

11. The method according to claim 10, wherein exons 6, 10, 17 and 18 are amplified using the primers identified by Sequence ID Nos. 11, 12, 19, 20, 59, 60, 61 and 62, or primers complementary thereto.

12. The method according to claim 3, wherein exons 4, 12 and 16 of the BRCA1 gene are amplified in one multiplex amplification reaction.

13. The method according to claim 12, wherein exons 4, 12, and 16 are amplified using the primers identified by Sequence ID Nos. 7, 8, 49, 50, 57 and 58, or primers complementary thereto.

14. The method according to claim 3, wherein exons 8, 13, 19 and 24 of the BRCA1 gene are amplified in one multiplex amplification reaction.

15. The method according to claim 14, wherein exons 8, 13, 19 and 24 are amplified using the primers identified by

Sequence ID Nos. 15, 16, 51, 52, 63, 64, 73 and 74, or primers complementary thereto.

16. The method according to claim 3, wherein exons 20, 22, and 23 of the BRCA1 gene are amplified in one multiplex amplification reaction.

17. The method according to claim 6, wherein exons 20, 22, and 23 are amplified using the primers identified by Sequence ID Nos. 65, 66, 69, 70, 71 and 72, or primers complementary thereto.

18. The method according to claim 3, wherein exons 2 and 20 are amplified in one multiplex amplification reaction.

19. The method according to claim 3, wherein the sequence is determined by amplifying a selected one of the multiplex amplification products in an aliquot of the multiplex reaction mixture and then sequencing the amplified multiplex amplification products.

20. The method according to claim 19, wherein the amplification of the multiplex reaction mixture and the sequencing of the amplified multiplex reaction product are performed in a single vessel.

21. The method according to claim 19, wherein the multiplex reaction mixture is amplified by combining the multiplex reaction mixture with an amplification mixture containing two primers for the selected one of the multiplex reaction products, a mixture of dNTP's and a thermostable polymerase in a buffer suitable for amplification, and exposing the resulting combination to a first series of thermal cycles including at least an extension phase and a denaturation phase to produce an amplified mixture containing the amplified multiplex reaction product; adding a sequencing mixture comprising a chain terminating nucleoside triphosphate to the amplified mixture and exposing the resulting combination to a second series of thermal cycles including at least an extension phase and a denaturation phase to produce sequencing fragments; and evaluating the size of the sequencing fragments.

22. The method according to claim 21, wherein the thermostable polymerase is Thermo Sequenase®.

23. The method according to claim 21, wherein the sequencing mixture further comprises a fluorescently labeled sequencing primer.

24. The method according to claim 23, wherein the sequencing mixture further comprises a thermostable polymerase

for sequencing which incorporates dideoxynucleosides into an extending oligonucleotide at a rate which is no less than about 0.4 times the rate of incorporation of deoxynucleosides in an amplification mixture.

25. The method according to claim 24, wherein the thermostable polymerase for sequencing is Thermo Sequenase®

26. A kit for testing a sample for mutations in the BRCA1 gene comprising a mixture of at least four oligonucleotide primers, said primers being selected to amplify at least two different exons or portions of exons of the BRCA1 gene in a multiplex amplification reaction.

27. The kit according to claim 26, wherein the primers are selected for amplification of exons 1 and 21 of the BRCA1 gene in one multiplex amplification reaction.

28. The kit according to claim 27, wherein the primers are for amplification of exons 1 and 21 are the primers identified by Sequence ID Nos. 1, 2, 69 and 70, or primers complementary thereto.

29. The kit according to claim 26, wherein the primers are selected for amplification of exons 2, 5, 9 and 14 of the BRCA1 gene in one multiplex amplification reaction.

30. The kit according to claim 29, wherein the primers for amplification of exons 2, 5, 9 and 14 are the primers identified by Sequence ID Nos. 3, 4, 9, 10, 17, 18, 55 and 56, or primers complementary thereto.

31. The kit according to claim 26, wherein the primers are selected for amplification of exons 3, 7 and 15 of the BRCA1 gene in one multiplex amplification reaction.

32. The kit according to claim 31, wherein the primers for amplification of exons 3, 7, and 15 are the primers identified by Sequence ID Nos. 5, 6, 13, 14, 57, and 58, or primers complementary thereto.

33. The kit according to claim 26, wherein the primers are selected for amplification of exons 6, 10, 17 and 18 of the BRCA1 gene in one multiplex amplification reaction.

34. The kit according to claim 33, wherein primers for amplification of exons 6, 10, 17 and 18 are the primers

identified by Sequence ID Nos. 11, 12, 19, 20, 61, 62, 63 and 64, or primers complementary thereto.

35. The kit according to claim 26, wherein the primers are selected for amplification of exons 4, 12 and 16 of the BRCA1 gene in one multiplex amplification reaction.

36. The kit according to claim 35, wherein the primers for amplification of exons 4, 12, and 16 are the primers identified by Sequence ID Nos. 7, 8, 51, 52, 59 and 60, or primers complementary thereto.

37. The kit according to claim 26, wherein the primers are selected for amplification of exons 8, 13, 19 and 24 of the BRCA1 gene in one multiplex amplification reaction.

38. The kit according to claim 37, wherein the primers for amplification of exons 8, 13, 19 and 24 are the primers identified by Sequence ID Nos. 15, 16, 53, 54, 65, 66, 75 and 76, or primers complementary thereto.

39. The kit according to claim 26, wherein the primers are selected for amplification of exons 20, 22, and 23 of the BRCA1 gene in one multiplex amplification reaction.

40. The kit according to claim 39, wherein the primers for amplification of exons 20, 22, and 23 are the primers identified by Sequence ID Nos. 67, 68, 71, 72, 73 and 74, or primers complementary thereto.

41. The kit according to claim 26, wherein the primers are selected for amplification of exons 2 and 20 in one multiplex amplification reaction.

42. The kit according to claim 41, wherein the primers for amplification of exons 2 and 20 are the primers identified by Sequence ID Nos.: 3, 4, 65 and 66.

43. An oligonucleotide primer having the sequence as set forth in any one of Sequence ID Nos. 1 through 77.

44. A method for testing a sample for mutations in the BRCA1 gene comprising the steps of

(a) amplifying one or more exons or partial exons of BRCA1 to produce amplified fragments;

(b) determining the sizes and amounts of the amplified fragments and comparing the determined sizes or amounts to standard values for amplification of the same exons or partial exons of wild-type BRCA1 gene, wherein a difference in fragment

size or amount is indicative of the presence of a mutation in the BRCA1 gene.

45. The method according to claim 44, wherein the amplification step is performed using two or more primers having the sequences as set forth Sequence ID Nos. 1 through 76.

46. The method according to claim 44, wherein two or more exons or partial exons are amplified in a multiplex amplification reaction to produce multiplex amplification products.

47. The method according to claim 46, further comprising the step of determining the sequence a selected one of the multiplex amplification products, wherein the sequence is determined by amplifying the selected one of the multiplex amplification products in an aliquot of the multiplex reaction mixture and then sequencing the amplified multiplex amplification products.

48. The method according to claim 47, wherein the multiplex reaction mixture is amplified by combining the multiplex reaction mixture with an amplification mixture containing two primers for the selected one of the multiplex

reaction products, a mixture of dNTP's and a thermostable polymerase in a buffer suitable for amplification, and exposing the resulting combination to a first series of thermal cycles including at least an extension phase and a denaturation phase to produce an amplified mixture containing the amplified multiplex reaction product; adding a sequencing mixture comprising a chain terminating nucleoside triphosphate to the amplified mixture and exposing the resulting combination to a second series of thermal cycles including at least an extension phase and a denaturation phase to produce sequencing fragments; and evaluating the size of the sequencing fragments.

49. The method according to claim 49, wherein the thermostable polymerase is Thermo Sequenase®.

50. The method according to claim 49, wherein the sequencing mixture further comprises a fluorescently labeled sequencing primer.

51. The method according to claim 51, wherein the sequencing mixture further comprises a thermostable polymerase for sequencing which incorporates dideoxynucleosides into an extending oligonucleotide at a rate which is no less than about 0.4 times the rate of incorporation of deoxynucleosides in an amplification mixture.

52. The method according to claim 52, wherein the thermostable polymerase for sequencing is Thermo Sequenase®

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